



## COL4A4 gene

collagen type IV alpha 4 chain

### Normal Function

The *COL4A4* gene provides instructions for making one component of type IV collagen, which is a flexible protein. Specifically, this gene makes the alpha4(IV) chain of type IV collagen. This chain combines with two other types of alpha (IV) chains (the alpha3 and alpha5 chains) to make a complete type IV collagen molecule. Type IV collagen molecules attach to each other to form complex protein networks. These networks make up a large portion of basement membranes, which are thin sheet-like structures that separate and support cells in many tissues. Type IV collagen alpha3-4-5 networks play an especially important role in the basement membranes of the kidney, inner ear, and eye.

### Health Conditions Related to Genetic Changes

#### Alport syndrome

More than 20 mutations in the *COL4A4* gene have been found to cause Alport syndrome. Most of these mutations change single protein building blocks (amino acids) in a region where the alpha4(IV) collagen chain combines with other type IV collagen chains. Other mutations in the *COL4A4* gene severely decrease or prevent the production of alpha4(IV) chains. As a result, there is a serious deficiency of the type IV collagen alpha3-4-5 network in the basement membranes of the kidney, inner ear, and eye. In the kidney, other types of collagen accumulate in the basement membranes, eventually leading to scarring of the kidneys and kidney failure. Mutations in this gene can also lead to abnormal function in the inner ear, resulting in hearing loss.

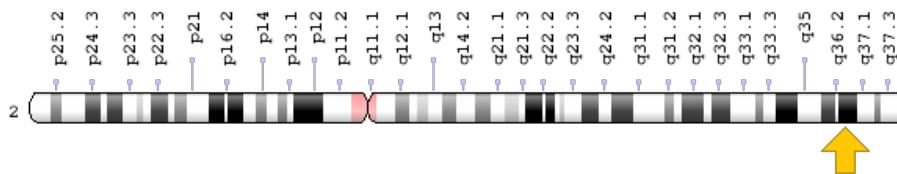
#### other disorders

Mutations in the *COL4A4* gene have been found to cause thin basement membrane nephropathy. This condition typically causes people to have blood in their urine (hematuria) but no other signs or symptoms of kidney disease. In the past, this condition was often called benign familial hematuria. Thin basement membrane nephropathy rarely progresses to kidney failure.

## Chromosomal Location

Cytogenetic Location: 2q36.3, which is the long (q) arm of chromosome 2 at position 36.3

Molecular Location: base pairs 226,970,293 to 227,164,559 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- alpha 4 type IV collagen
- CA44
- CO4A4\_HUMAN
- Collagen IV, alpha-4 polypeptide
- collagen of basement membrane, alpha-4 chain
- collagen type IV alpha 4
- collagen, type IV, alpha 4

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): A model of the molecular structure of a basal lamina  
<https://www.ncbi.nlm.nih.gov/books/NBK26810/?rendertype=figure&id=A3581>
- Molecular Biology of the Cell (fourth edition, 2002): Basal Laminae Perform Diverse Functions  
<https://www.ncbi.nlm.nih.gov/books/NBK26810/#A3583>

### GeneReviews

- Alport Syndrome and Thin Basement Membrane Nephropathy  
<https://www.ncbi.nlm.nih.gov/books/NBK1207>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28COL4A4%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

## OMIM

- COLLAGEN, TYPE IV, ALPHA-4  
<http://omim.org/entry/120131>
- HEMATURIA, BENIGN FAMILIAL  
<http://omim.org/entry/141200>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_COL4A4.html](http://atlasgeneticsoncology.org/Genes/GC_COL4A4.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=COL4A4%5Bgene%5D>
- HGNC Gene Family: Collagens  
<http://www.genenames.org/cgi-bin/genefamilies/set/490>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=2206](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2206)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/1286>
- UniProt  
<http://www.uniprot.org/uniprot/P53420>

## **Sources for This Summary**

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